Helping to Improve Human Identity Testing: Development of 26 New miniSTR Loci as DNA Markers

NIST is developing a comprehensive set of human identity DNA markers to enable more accurate DNA analysis for human identity testing. Based upon sequences reported in the literature, 26 new miniature short tandem repeats (miniSTRs) were designed to maximize their utility for human identity testing. The sequences of the new miniSTR are now available on the NIST website. The 26 new miniSTRs are being calibrated for use in tandem with the widely used NIST SRM 2391b to enable the highest integrity human DNA identity testing.

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Genetic tests that target smaller sections of DNA are more successful at recovering information from highly degraded biological specimens. The value of miniature short tandem repeat (miniSTR) assays were demonstrated during efforts to identify the 9/11 World Trade Center victims. Additional loci are being developed at NIST to help expand the capabilities of miniSTR testing.

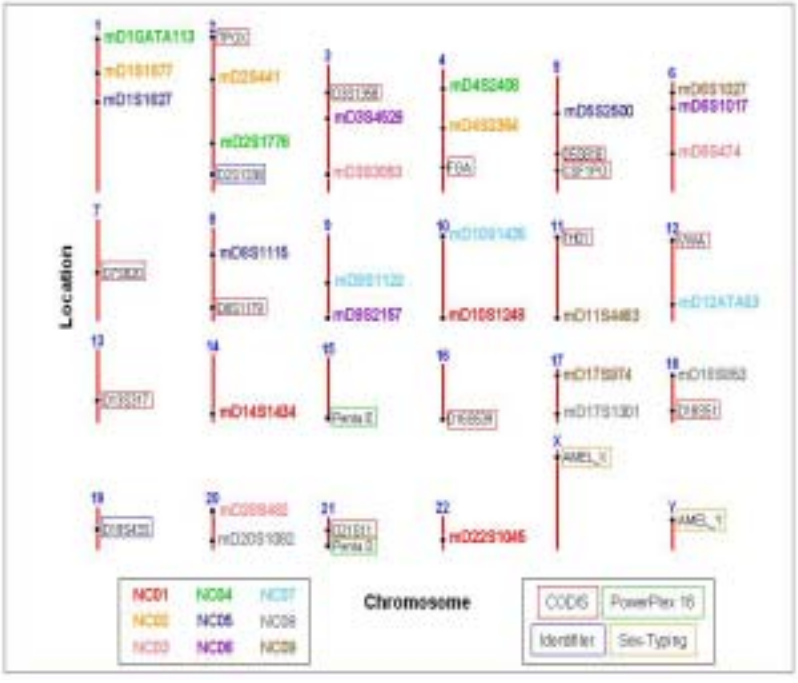
A number of studies have demonstrated that successful analysis of degraded DNA specimens from mass disasters or forensic evidence improves with smaller sized polymerase chain reaction (PCR) products. If DNA is exposed to the elements or to fire for any length of time, degradation can occur due to bacterial, biochemical or oxidative processes. Within the forensic community, a core set of short tandem repeat (STR) markers have been developed for utilization in forensic casework, and large DNA databases such as the Combined DNA Index System (CODIS) have been developed incorporating these markers.

An initial effort to reduce the STR amplicon size for CODIS loci resulted in a set of miniplexes to analyze degraded DNA. However, new autosomal STR loci are being examined because many of the CODIS core loci contain repeat flanking regions that are not amenable for redesigned primers (e.g. D7S820) or have large allele ranges (e.g., D21S11 and FGA) that make it impossible to create small PCR products. We have therefore scanned the literature for new STR loci and designed and optimized new PCR primers to reduce the amplicon size as much as possible. The loci were evaluated for variability in a set of more than 600 U.S. population samples, and selected alleles were sequenced in order to calibrate the allele nomenclature. The 26 chosen loci contain alleles that are less than 150 bp in size and are well-spaced across the human genome. The candidate loci are all either located on chromosomes that differ from the 13 CODIS core loci or are at least ~ 50 Mb apart from an existing CODIS loci on the same chromosome, and therefore unlinked from that particular marker (see figure for their exact locations in relation to the CODIS loci). In addition, by moving PCR primers closer to the STR region, we have established that it is possible to decrease the chance of allele or locus-dropout that may occur in degraded samples. In fact, the value of these new loci have been confirmed in comparing the success of the miniSTR assays for typing degraded bone samples and aged blood and saliva stains while partial profiles were observed with the majority of the samples using a commercial STR kit.

A portion of the NIST STRBase website is being devoted to information on the 26 new miniSTR loci: http://www.cstl.nist.gov/biotech/strbase/newSTRs.htm.

Some of our first miniSTR loci characterized--D2S441, D10S1248, and D22S1045--have been selected by the European community as recommended STRs for adding to their core genetic systems used for human identity testing.

The figure illustrates the chromosomal locations of the miniSTR markers in relation to the previously established CODIS markers



A major commercial manufacturer, Applied Biosystems, is currently developing a new miniSTR kit based in large measure on our pioneering work here at NIST. We are also in the process of certifying the genotype values for these 26 newly characterized loci in the genomic DNA components of the widely used NIST SRM 2391b to enable calibration of future measurements made with these loci by laboratories around the world.

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